



## Under-5 mortality and the contribution of congenital disorders in South Africa

**To the Editor:** The article 'Child mortality in South Africa: Fewer deaths but better data are needed',<sup>[1]</sup> which appeared in the March 2018 *SAMJ* Maternal and Child Health Supplement, reviews nationally representative mortality data and the causes of death in children under 5 years of age in South Africa (SA). Two different terms are used synonymously without definition in this article – 'congenital disorders' and 'congenital abnormalities'. By definition, congenital disorders (CDs) are abnormalities in structure or function present from birth, including inborn errors of metabolism.<sup>[2]</sup> Congenital abnormalities (often used interchangeably with 'congenital anomalies') are a subset of CDs, previously defined in the literature as obvious structural abnormalities as categorised in Chapter XVII: 'Congenital malformations, deformations and chromosomal abnormalities' in the *International Statistical Classification of Diseases and Related Health Problems* (ICD-10) – which excludes a third of CDs included elsewhere in the ICD-10.<sup>[3-6]</sup>

Congenital abnormalities are also referred to by Bamford *et al.*<sup>[1]</sup> as a separate entity from non-communicable diseases (NCDs), contrary to the World Health Organization (WHO) definition of CDs as an NCD and the concept of CDs as being the first NCD experienced in life.<sup>[7-10]</sup> Excluding CDs from this disease category has implications for achieving the Sustainable Development Goal (SDG) 3 target of reducing premature mortality from NCDs by one-third.<sup>[11]</sup>

While the abovementioned article highlights potential underestimates in the vital registration data of deaths due to HIV infection and malnutrition, this consideration is not given for CD diagnoses omitted from death certificates.<sup>[1]</sup> This is despite the parallel between these diagnoses in being omitted as the correct recorded cause of death (contributory or otherwise) due to reluctance with regard to HIV cases and non-/misdiagnosis of CDs. The Child Problem Identification Programme (Child PIP), in particular, has cited a lack of clinicians necessary to make accurate diagnoses as a limitation in the cause of death categorisation.<sup>[12,13]</sup>

The recommendation by Bamford *et al.*<sup>[1]</sup> to scale up interventions to reduce deaths from emerging causes of under-5 child death includes congenital abnormalities, which have risen from 3.9% in 2011 as a proportional cause of death to 5.9% in 2015. Combined with the simultaneous reduction in infectious diseases, e.g. diarrhoea and pneumonia, this is indicative of the ongoing positive epidemiological transition in SA.<sup>[9,14]</sup> Following the trend of high-income countries where CDs account for up to 28% of under-5 deaths,<sup>[15]</sup> it is imperative that these interventions include relevant medical genetic services for the care and prevention of those affected by and at risk of CDs. With up to 70% of CDs potentially prevented or cured and subsequent disability ameliorated, the potential impact of addressing CDs cannot be ignored owing to the sizeable proportion of preventable child deaths.<sup>[16-19]</sup> This makes the prioritisation of CDs key to reducing preventable child and neonatal deaths and to attaining the SDG targets.<sup>[11]</sup>

For the quality of data in death notification forms to improve, capacity must be increased to ensure the accurate diagnosis of CDs as the cause of death for both neonatal and child deaths. With tertiary-level genetic services available in only 3 of the 9 provinces and national surveillance grossly underreporting CDs, there is much room for improvement.<sup>[20,21]</sup> It is anticipated that the restructuring of disease surveillance, including CDs, under the National Public Health Institute of South Africa (NAPHISA) will rectify some of these issues, but more targeted action is required.<sup>[22]</sup>

To achieve the SDG 3 target to reduce the SA under-5 mortality rate from 37 - 40 deaths per 1 000 live births to the required target of <25 per 1 000, a 37% reduction is required.<sup>[1,11]</sup> Previously unprioritised health issues, including CDs, must now be addressed for this target to be reached. The literature is clear on the role of comprehensive medical genetic services in significantly reducing child mortality. With an infant mortality rate of 27 - 33 deaths per 1 000 live births, SA is long past the point of 40 infant deaths per 1 000 live births, when countries should implement these services.<sup>[1,8,10,16,23]</sup> To meet SDG 3 by 2030, the role of CDs in neonatal, infant and child mortality must be comprehensively addressed as a priority in SA.

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